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(21) International Application Number: PCT/CA99/00646 (22) International Filing Date: 20 July 1999 (20.07.99) (30) Priority Data: 60/093,495 20 July 1998 (20.07.98) US 60/130,269 21 April 1999 (21.04.99) US (71)(72) Applicants and Inventors: SCHERER, Stephen, W. [CA/CA]; 60 Mayfield Avenue, Toronto, Ontario M6S 1K5 (CA). MINASSIAN, Berge, A. [CA/CA]; 18 Vanderhoof Avenue, Toronto, Ontario M4G 2H1 (CA). DELGADO-ESCUETA, Antonio [US/US]; 28342 Rey De Copas Lane, Malibu, CA 90265 (US). ROULEAU, Guy [CA/CA]; 4850 Cote St. Luc Road, Montreal, Quebec H3W 2H2 (CA). (74) Agent: BERESKIN & PARR; 40th Floor, 40 King Street West, Toronto, Ontario M5H 3Y2 (CA).		(81) Designated States: AE, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CU, CZ, DE, DK, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, UA, UG, US, UZ, VN, YU, ZA, ZW, ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG). Published <i>With international search report.</i> (88) Date of publication of the international search report: 11 May 2000 (11.05.00)	
(54) Title: LAFORA'S DISEASE GENE			
(57) Abstract A novel gene (EPM2A) that is deleted or mutated in people with Lafora's disease is described. The EPM2A gene encodes a protein having an active catalytic site of a protein tyrosine phosphatase. Many different sequence mutations as well as several microdeletions in EPM2A have been found that co-segregate with Lafora's disease.			

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INTERNATIONAL SEARCH REPORT

Inter. Appl. No.

PCT/CA 99/00646

A. CLASSIFICATION OF SUBJECT MATTER

IPC 7 C12Q1/68 C12N9/16 C12N15/55 C07K14/47

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	EMBL Database Entry HS466P17 Accession number AL023806; 8 June 1998 Thorpe K.: "Human DNA sequence from clone 466P17 on chromosome 6q24: Contains a putative novel gene, the 5' part of the EPM2A (Laforin) gene...and a ca repeat polymorphism". XP002127924 see database entry	1-9, 17-19
P, X	MINASSIAN B ET AL: "Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy" NATURE GENETICS, vol. 20, no. 2, October 1998 (1998-10), pages 171-74, XP000869599 the whole document	1-19
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Date of the actual completion of the international search

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INTERNATIONAL SEARCH REPORT

International Application No

PCT/CA 99/00646

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
P, X	SERRATOSA J ET AL: "A novel protein tyrosine phosphatase gene is mutated in progressive myoclonus epilepsy of the Lafora type (EPM2)" HUMAN MOLECULAR GENETICS, vol. 8, no. 2, February 1999 (1999-02), pages 345-52, XP000867126 the whole document	1-19
A	MINASSIAN B ET AL : "Progresss towards the positional cloning of a gene for Lafora's disease" NEUROLOGY , vol. 48, no. 3 suppl 2, 12 - 19 April 1997, page A428 XP000866505 see abstract P06.091	1-19
A	SAINZ J ET AL: "Lofora's progressive myoclonus epilepsy : narrowing the chromosome 6Q24 locus by recombinations and homozygosities" AMERICAN JOURNAL OF HUMAN GENETICS, vol. 61, no. 5, 1997, pages 1205-09, XP000866531 figure 1	1-19